Chronic Disease Prevention and Genomics
2004 Activity Snapshot

The National Center for Chronic Disease Prevention and Health Promotion (NCCDPHP) continues to look at the important links between chronic disease and genetics. Augmenting the work of the Office of Genomics and Disease Prevention, NCCDPHP is growing the science around some of the leading causes of death – diseases such as heart disease, cancer, and diabetes. Today, we know that many chronic diseases are preventable and will become more fully understood only when both the genetic and environmental contributions to their etiology are known. CDC is committed to advancing the science to improve health and prevent and control chronic diseases. The following document highlights CDC supported chronic disease/genomics activities, including epidemiologic studies, planning and policy activities, and dissemination and education efforts.

2004 Major Accomplishments

Exploring the Links between Chronic Diseases and Family History

One of the primary activities, a collaborative effort of OGDP and NCCDPHP, is the Family History Public Health Initiative, which began in 2002 with an extensive literature review and a subsequent paper that examined the idea of using family history for disease prevention. NCCDPHP participates in OGDP's Family History Work Group, which was formed to conduct research and develop a family history tool for disease prevention. The multidisciplinary group has established criteria for diseases to include in a family history tool, reviewed the literature for nearly 40 diseases, and developed a prototype tool with 16 diseases. Ongoing work includes pilot studies to further refine the tool, development of algorithms to assess risk, development of a resource manual for primary care providers, and design and funding of studies to evaluate the validity and utility of the approach.

Family History of Cancer and Cancer Screening

Little data are available on the use of screening tests among individuals with a family history of cancer. Using data from the 2000 National Health Interview Survey (NHIS), a logistic regression model is being used to examine the association between first-degree family history of cancer and receipt of a recent screening test (mammography, prostate specific antigen, and colonoscopy or flexible sigmoidoscopy) among persons who had at least one screening test, adjusting for age, race, gender, education, income, marital status, and health insurance coverage.

Genetic Testing for Breast and Ovarian Cancer Susceptibility: Evaluating Direct-to-Consumer Marketing

Breast and ovarian cancer are the second and fifth leading causes of cancer death, respectively, among women in the United States. Mutations in two genes, BRCA1 and BRCA2 (BRCA1/2), are associated with predisposition for inherited breast and ovarian cancer and are identified in 5% to 10% of women with breast or ovarian cancer (BOC). Since 1996, genetic testing for these mutations has been available clinically; however, population-based screening is not recommended because of the complexity of test interpretation and limited data on clinical validity and utility. Despite the test's limited applicability in the general population, the U.S. provider of clinical BRCA1/2 testing (Myriad Genetic Laboratories, Inc.,

Salt Lake City, Utah) conducted a pilot direct-to-consumer (DTC) marketing campaign in two cities (Atlanta, Georgia, and Denver, Colorado) during September 2002–February 2003. Although DTC advertisements have been used to raise consumer awareness about pharmaceuticals, this was the first time an established genetic test was marketed to the public. To assess the impact of the campaign on consumer behaviors and healthcare provider practices, CDC and the respective state health departments for the pilot cities and two comparison cities (Raleigh-Durham, North Carolina, and Seattle, Washington) surveyed consumers and providers. This report summarizes results of those surveys, which indicated that consumer and provider awareness of BRCA1/2 testing increased in the pilot cities and that providers in these cities perceived an impact on their practice (e.g., more questions asked about testing, more BRCA1/2 tests requested, and more tests ordered). However, in all four cities, providers often lacked knowledge to talk to patients about inherited BOC and testing. These findings underscore the need for evidence-based recommendations on appropriate use of genetic tests and education of providers and the public to achieve maximum individual and public health benefit from genetic testing. This project is a collaboration between NCCDPHP and OGDP.

Genomics and Chronic Disease: What Project Officers Need to Know
CDC, in conjunction with the Universities of Michigan, North Carolina, and Washington, presented "Genomics and Chronic Disease: What Project Officers Need to Know" in June 2004. This workshop was designed to provide an introduction to genomics to NCCDPHP project officers and help them better understand how existing chronic disease programs – particularly in the areas of diabetes and cancer – have incorporated genomics into their existing programs.

Hereditary Hemochromatosis
CDC and its partners continue to support an Internet-based teaching module on Hereditary Hemochromatosis for health professionals and the public, including the development and dissemination of education materials, which include the patient brochure *Iron Overload and Hemachromatosis: Information for Patients and Family.*

Johnston County Osteoarthritis Project
This ongoing community-based cohort study of rural white and black persons to determine the prevalence, incidence, and factors associated with the occurrence or progression of hip and knee osteoarthritis includes a genomics component for examining genes (HH, COMP, COL2A, others to be determined) that may be linked to osteoarthritis and related conditions. The study is conducted by the University of North Carolina with CDC funding and collaboration.

Oregon Sudden Unexplained Death Study
Despite current medical knowledge, sudden cardiac death – or the sudden failure of heart rhythm – is unexplained in up to 15 percent of cases. The Oregon Sudden Unexplained Death Study (SUDS) Group, funded by CDC, is conducting research on the mechanisms of sudden cardiac death. This prospective study of all patients suffering sudden cardiac death in Multnomah County, Oregon, will develop a comprehensive database describing patients' medical histories, including clinical findings, pathologic data, and genetic analysis.

State Capacity Grants for Integrating Genomics into Chronic Disease Prevention Programs
State and community health agencies recognize the need to augment existing genetics expertise in maternal and child health and newborn screening capacity to integrate genomics into a wider range of disease control and prevention programs. To develop this capacity, NCCDPHP established cooperative agreements with state health departments in Michigan, Minnesota, Oregon, and Utah to strengthen

leadership and promote coordination in programs for genomics and chronic disease prevention. Through these agreements, those states are working on a variety of projects over a five year period from 2003 to 2008. These projects include integrating genomics and family history into ongoing and new population-based strategies for identifying and reducing the burden of specific chronic, infectious, and other diseases; enhancing planning and coordination for integrating genomics into core state public health specialties (such as epidemiology, laboratory activities, and environmental health); and facilitating the use of family history and new knowledge about gene-environment interactions to enhance chronic disease prevention. This project is a collaboration between NCCDPHP and OGDP.

Stroke Prevention in Young Women
A population-based case control study examining gene-environment interactions in the risk of stroke, the study will determine the prevalence of genetic polymorphisms in the population, gene-environment interactions in stroke risk. The study also will determine the population-attributable fraction associated with genetic risk factors and gene-environment interaction. The study is conducted in collaboration with University of Maryland.

Future Directions: Opportunities Ahead for Genomics and Chronic Disease Prevention

In 2005 and beyond, NCCDPHP will continue its work in genomics in the following priorities areas:

- Foster efforts to use family history to promote health – particularly through support and expansion of CDC's family history project.
- Support state based grants to build capacity in states to understand the links and importance of family history in chronic disease prevention and health promotion.
- Promote communication and policy efforts to foster the public's understanding of genomics and the role in health promotion.